

Current concepts in physiotherapy and rehabilitation protocol for arthrogryposis multiplex congenita: An unusual case report

Physiotherapy and rehabilitation protocol of arthrogryposis multiplex congenita

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Abstract

Arthrogryposis multiplex congenita is a non-progressive syndrome characterized by multiple congenital joint contractures. This case is a six-year-old child with arthrogryposis multiplex congenita who presented at Inonu University Orthopedic Clinic with complaints of weakness and atrophy of m. flexor and extensor carpi radialis and ulnaris, m. flexor and extensor digitorum superficialis, biceps, triceps muscles in the bilateral arms. The tendon transfer was performed in the attachment of the triceps muscle due to the absence of brachialis. This case report presents information on differential diagnosis, detailed symptoms, and physical therapy management of a six-year-old patient with arthrogryposis multiplex congenita.

An early approach to physical therapy decreases complications such as flexor contracture of the wrist and elbow, muscle weakness and atrophy following arthrogryposis multiplex congenita. Physical therapy interventions can improve rigid contracture and muscle atrophy and prolong the need for surgical operation in childhood.

Keywords

Physical Rehabilitation; Arthrogryposis; Atrophy; Children

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Introduction

Arthrogryposis multiplex congenita (AMC) is a non-progressive syndrome characterized by multiple congenital joint contractures [1]. It depends on a multifactorial etiology, and there are autosomal recessive, autosomal dominant, X-linked (more observed in males) or mitochondrial inheritance. 1/3000 of the detected arthrogryposis multiplex congenita is also called amyoplasia congenita or multiple congenital articular rigidity. It may occur due to joint limitation during fetal development because of the mother's narrow uterus. The disease is divided into two main types due to neurological or myopathic disorders, and neurological symptoms are observed in most patients. Also, in children diagnosed with AMC, the incidence of scoliosis is between 30% and 67% in childhood and the curves generally progress by 6.5° per year [1, 2]. AMC occurs because of intrinsic or fetal anomalies and extrinsic causes. As fetal abnormalities, congenital myopathy and muscular dystrophies are noticed on account of connective tissue disorders such as amyoplasia, peripheral or central nervous system anomalies, diastrophic dysplasia, connective tissue diseases, ligamentous and bone anomalies, dermatopathies, osteochondroplasia. Extrinsic causes may occur owing to maternal infection (rubella, Coxsackie), uterine abnormalities or tumours, multiple pregnancies, intrauterine vascular problems and traumas [2]. In AMC cases, the extremities are generally weakened, and the muscles appear fusiform. Sensory perception is normal, but a decrease or loss of the deep tendon reflex is observed. [3]. Amyoplasia is the most common form of AMC and constitutes 1/3 of all cases. Symmetrical contractures are usually detected in 85% of children diagnosed with amyoplasia, and generally, all extremities are affected. Very few of them have genital abnormalities, congenital hernia, amniotic band, congenital heart disease and respiratory problems. Generally, in AMC cases, internal rotation, adduction in the shoulders, extension in the elbow, and flexion position in the wrist are observed. There are internal rotation, adduction in the shoulder, extension in the elbow and flexor contracture in the wrist. In severe cases, the hand may be fully flexed, and the finger joints in rigid flexion [2, 3]. Rigid flexor contracture in the interphalangeal joint was not detected in this case. Dysplasia of the hip joint of the affected lower extremity is often observed. The leg joints are more affected than the arm joints. The affected extremities are soft tube-shaped and hypoplastic [4]. However, in this case, the lower extremity has completed its normal development, and walking symmetry, speed, cadence and duration are expected. Other joints involved are elbow, knee, wrist, ankle, fingers and hip joints. In this case, rigid elbow extension contracture and flexion limitation due to arthrodesis in the elbow joint were observed. The objects given due to 60° flexor contractures in the right wrist cannot be taken bilaterally and brought to the opposite shoulder or mouth.

Based on the ICF (International Classification of Functioning, Disability and Health) criteria and treatment guidelines for physiotherapists, physical therapy management for patients with AMC may include gentle joint manipulation and massage application consisting of stroking, kneading, picking up and rolling applications, removable splint management for knees and feet to help allow regular muscle movement [4]. Also,

orthosis management can aid walking and independence for children with AMC [3, 4], by maintaining muscle tension and strengthening the patient's muscles, especially the hip extensors, quadriceps and deltoid anterior, middle, and posterior part, rotator cuff muscles, trapezius and serratus anterior muscles [2, 4]. It helps to encourage active muscle use to prevent immobilization, stretching of joint and muscle contractures, and initiates a neck, pectoral, elbow, wrist and finger flexor muscle stretching program for the family to do at home. It is recommended to stretch 3-5 times a day, 3-5 repetitions per set, holding each stretch for 20-30 seconds [5]. Aquatherapy, hippotherapy, training in the use of assistive devices such as a gait instructor, walker, crutches, orthosis, dynamic strengthening of the trunk, ambulation independently or with an assistive device are other treatment methods in the physical therapy for children with AMC [4, 5].

This study aimed to provide information about the duration and frequency of physical therapy to be applied to children with AMC in play age and create an appropriate rehabilitation protocol to provide information on its content.

Case Report

To conduct this study, informed consent was obtained from the patients' family. In this case, anencephaly, meningomyelocele, and holoprosencephaly were not observed. There was no fetal immobility due to these reasons. Besides, the mother had no history of multiple pregnancy or oligohydramnios. There was no central nervous system (CNS) anomaly in this case, and the tendon reflex was normal. Cognition and mental health were in line with their peers. Joint stiffness, column vertebral, and flexor contracture in the distal and proximal joints were detected. Hypoplasia and aplasia were noticed in m. flexor and extensor carpi radialis and ulnaris, m. flexor and extensor digitorum superficialis, biceps, triceps and deltoid muscles. Lower extremity muscle tone, which was evaluated with resistance during passive motion, was expected in the range of motion. Adduction and internal rotation in the shoulders, forearm pronation, flexion contracture on the left and right sides of the wrist and ulnar deviation were observed. There was no hip dislocation, unilateral or bilateral hip flexion contracture in the lower extremity. Extension contracture of the knees or standing pes calcaneus, talipes equinovarus was not observed. Blood glucose, urea and electrode levels were normal.

Description of the Evaluation Outcomes

This case was six years old, and there was an active contraction of the right flexor carpi radialis. The tendon transfer was performed in the attachment of the triceps muscle due to the absence of brachialis. Shoulder flexion was bilaterally 70°, shoulder abduction 60°, and tends to compensate, especially shoulder flexion with neck flexion (Figure 1). Elbow flexion was limited at 90°, wrists were restrained in extension at 20°, and both hips and knees had normal joint movement in all directions. In short, a decrease in range of motion was observed in the extremities and neck. Throughout the passive movement, there was an increase in pain, asymmetrical posture, a reduction in comfort in the prone position, difficulties in participating in the social environment, and a decrease in the movements required for daily life activities and playing games. The cylindrical grip

fails due to flexor contracture in the wrist. There was a decrease in the fingers' muscle strength, and the elbow's skin piles have disappeared. Both elbows have webbing. Diaphragmatic dwarfism was present. Also, there was hypoplasia in the pectoral muscles (Figure 2). Abdominal breathing was dominant in the patient, and chest respiration was restricted. Mental retardation was not detected in our patient. Cleft lip and palate were not observed.

Sitting balance was full, and kyphosis occurred in sitting and standing position. Muscle strength (according to the Modified Oxford Scale): Global 3-4 / 5 in the upper extremities, global 5/5 in the lower extremities, and walking was natural. The evaluation made according to the ICF model is given in Table 1.

Description of Intervention

The AMC treatment in this study aims to increase the passive range of motion and strengthen active movement. The experimental stage has been reached by stabilizing the joints and loosening the contractures with surgery. If there were flexion or extension contractures in the shoulders and elbows, stretches were given to reduce this. Passive exercises, bandaging, and corrective plastering were essential for the treatment. Wrist flexor contracture and thumb adductor orientation were reduced with serial casts applied for six weeks. As a superficial heating agent in joints with contractures before exercise, a hot pack for 15 minutes, and as a deep heating agent to increase connective tissue elasticity in joints with arthrodesis, ultrasound (1,5 w/cm²) was applied for 5 minutes. It is possible to open contractures with thermoplastic splints. A wrist flexor contracture with a night splint was used for 6-9 weeks. The wrist was left in 20° extension, the metacarpophalangeal joint at 25° flexion, and in semiflexion due to absence of flexor contracture in the fingers. Besides, since the abdominal and pectoral muscles were weak, pulmonary exercises combined with aerobic exercise were studied. Strengthening exercises were performed with active assistive and resistive (theraband, 0.5 kg or 1 kg dumbbells, sandbags) exercises. Additionally, electrical stimulation was given to the relevant muscles for 15 minutes in the strengthening program. The exercise program applied during these 12 weeks was done at least 3-4 times a week, 3-5 times a day, 45 minutes, 20-30 repetitions of each movement, and the splint was attached immediately thereafter to preserve the obtained angle.

To treat the wrist contracture, both wrists were stretched, and the sensory input was provided by approaching the shoulder joint capsule by weight transfer. To prevent kyphotic posture, prone positioning on the hands and elbows was recommended for most of the day, except for one hour after each meal. Pectoral stretches and back muscles stimulation were performed in the supine position on the physioball.

In our case patient, after rehabilitation, an increase in the range of motion of the upper extremity (Table 2), the muscle strength measured before physical therapy (according to the Modified Oxford Scale) was generally 3-4 / 5 in the upper extremities, and 5/5 in the lower extremities, after treatment, the upper and lower extremities were 5/5 when measuring; Muscle tone, which was evaluated with resistance during passive movement, was found to be expected in the range of motion. Pinching grip strength was measured as 2.33 before treatment on the right



Figure 1. Limitation of shoulder flexion and exposure to a compensated range of motion



Figure 2. The atrophy of pectoral muscle, arm, and forearm muscle

Table 1. Evaluation criteria according to the ICF model

Health State		
Arthrogryposis Multiplex Congenita Disease		
↓		
Body Function / Structural Inadequacy	Activity Limitation	Restricted Participation
Decreased limb and neck ROM Asymmetric posture Pain caused by passive movement	The decrease in movements required for daily activities and playing games Decrease in comfort in the prone position	Decreased communication with family and community and participation in social activities

Table 2. Results of measuring the range of motion of the upper limbs (ROM) before and after rehabilitation

Joints of the body	Movement	Pre-Rehab Right	Pre-Rehab Left	Post-Rehab Right	Post-Rehab Left
Shoulder	Flexion	70°	70°	120°	120°
	Extension	30°	30°	45°	45°
	Abduction	60°	60°	110°	110°
	Internal Rotation	70°	70°	70°	70°
	External Rotation	60°	60°	80°	80°
Elbow	Flexion	90°	90°	120°	120°
Wrist	Flexion	60°	90°	90°	90°
	Extension	20°	20°	60°	60°
Finger	Flexion	30°	30°	45°	45°

side, 3.66 after treatment, and 1.33 before treatment on the left side, and 2.66 after treatment. While the coarse grip was 9.33 before the treatment in the right hand, it was measured as 10.66 after the treatment; It was 8.66 before treatment in the left hand, and 9.66 after treatment. After rehabilitation, our patient has become able to perform fine motor skills such as holding objects, holding a cylinder and palmar, releasing an object, and button fastening. With the stretching of the pectoral muscle, the increase in back extensor and trapezius muscle strength, kyphotic posture was prevented and sitting in an upright position was achieved.

Discussion

Arthrogryposis multiplex congenita is a rare and heterogeneous disease. Children diagnosed with AMC should be handled with an interdisciplinary problem-solving approach. It has been found that there is little literature on rehabilitation strategies for treating children with AMC, which makes rehabilitation professionals more anxious when treating children with AMC. This case report provides information on rehabilitation sessions and specific treatment strategies for a growing play-age child with AMC.

Physical therapy for children with AMC plays an essential role in their independence. A recent review of AMC studies shows that the detailed description of a specific therapy session is not clearly stated in any literature. In this report, we tried to convey the duration, frequency (frequency) and treatment methods used in the treatment of a play-age boy with AMC.

Studies have been recommended for patients with AMC diagnosis, physical therapy and occupational therapy to increase postural muscle strength and maximal mobilization

and the motor ability for age-appropriate activities. If a patient diagnosed with AMC cannot hold objects, cannot provide personal care, and has eating problems, compensated devices (splints, adaptive equipment) are required. As soon as infants diagnosed with AMC are born, passive ROM is used to assess muscle strength by muscle contraction palpation, functional mobility and motor evaluations, the Alberta Infant Motor Scale, and The Bayley Scale Infant Developmental III or Peabody Developmental Motor Scale-II (PDMS-2). The PDMS-2 scale is used to assess fine and gross motor skills in newborn-71-month-old children.

In the study conducted by Azbell et al., they evaluated the efficacy of serial casting and physical therapy applied for clubfoot in the lower extremity in a 9-month-old patient with arthrogryposis multiplex congenita, whose initial evaluation was 11 days. Hip dysplasia was suspected due to the 5°-10° restriction as a result of hip abduction with the hip flexed at 90°. Passive ROM (shoulder flexion, abduction, shoulder internal and external rotation, elbow, wrist, finger flexion and extension, hip, knee extension and flexion, and ankle flexion), muscle strength assessment, FLACC pain scale was used to assess the amount of pain during passive motion and after ROM in the 4-5 minutes. It was aimed to reduce knee and hip flexor contracture with two repetitions of 30-second stretching on each contracted joint, neck stretching, night splint for the wrist, a dynamic splint for elbow, prone positioning [6]. In the cases reported by Azbell et al., elbow flexion restriction was due to extension contracture. They also used Mitchell shoes for clubfoot treatment [6]. Since our case had tendon transfer due to the absence of brachialis, active 90° elbow flexion took place. We preferred to use a dynamic splint to maintain active elbow flexion. Besides, in our study, family training was provided as a physical therapy practice to slow down the progression by stretching, muscle strengthening, motor ability facilitation, reduction in pain that restricts daily activity and using a splint in play, and prevention of secondary scoliosis. However, the locomotion abilities in PDMS-2 (Peabody Developmental Motor Scale) were not examined because our case was an ambulatory patient.

Patel et al. published a case report in which they shared the duration, frequency and effects of physical therapy for a girl with AMC in infancy, starting from 15 days and continuing until the age of 11 months. In evaluations using the ICF model, they have ensured to increase ROM patency in physical therapy, to preserve muscle tension, and to improve neuro-sensory motor development [7]. In our case study, the evaluation was made according to the ICF model. Fine and gross motor development was contributed by physical therapy, range of motion, muscle tension, and neuro-sensory motor facilitation.

Shinde et al. observed in their case study that a neurological and myopathic abnormality caused AMC. The neurological form is more common than the myopathic form. AMC’s symptoms are micrognathia, wide flat nose, low ear, high arched plaque, and hypoplastic diaphragm/lungs [8]. In our case, hypoplastic diaphragm/lung and atrophic pectoral muscles were observed as symptoms.

Conclusion

An early physical therapy approach reduces complications

such as arthrogryposis multiplex delivery, elbow extension contracture, wrist flexor contracture, muscle weakness and atrophy. It has an important place in increasing the range of motion of the upper extremity.

Physical therapy interventions can help improve rigid contracture and muscle atrophy and prolong the need for surgical operations in children's younger ages.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

Conflict of interest

None of the authors received any type of financial support that could be considered potential conflict of interest regarding the manuscript or its submission.

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